NEWBORN SCREENING ADVISORY COMMITTEE

October 1, 2019

NORTH Dakota | Health
Be Legendary.™

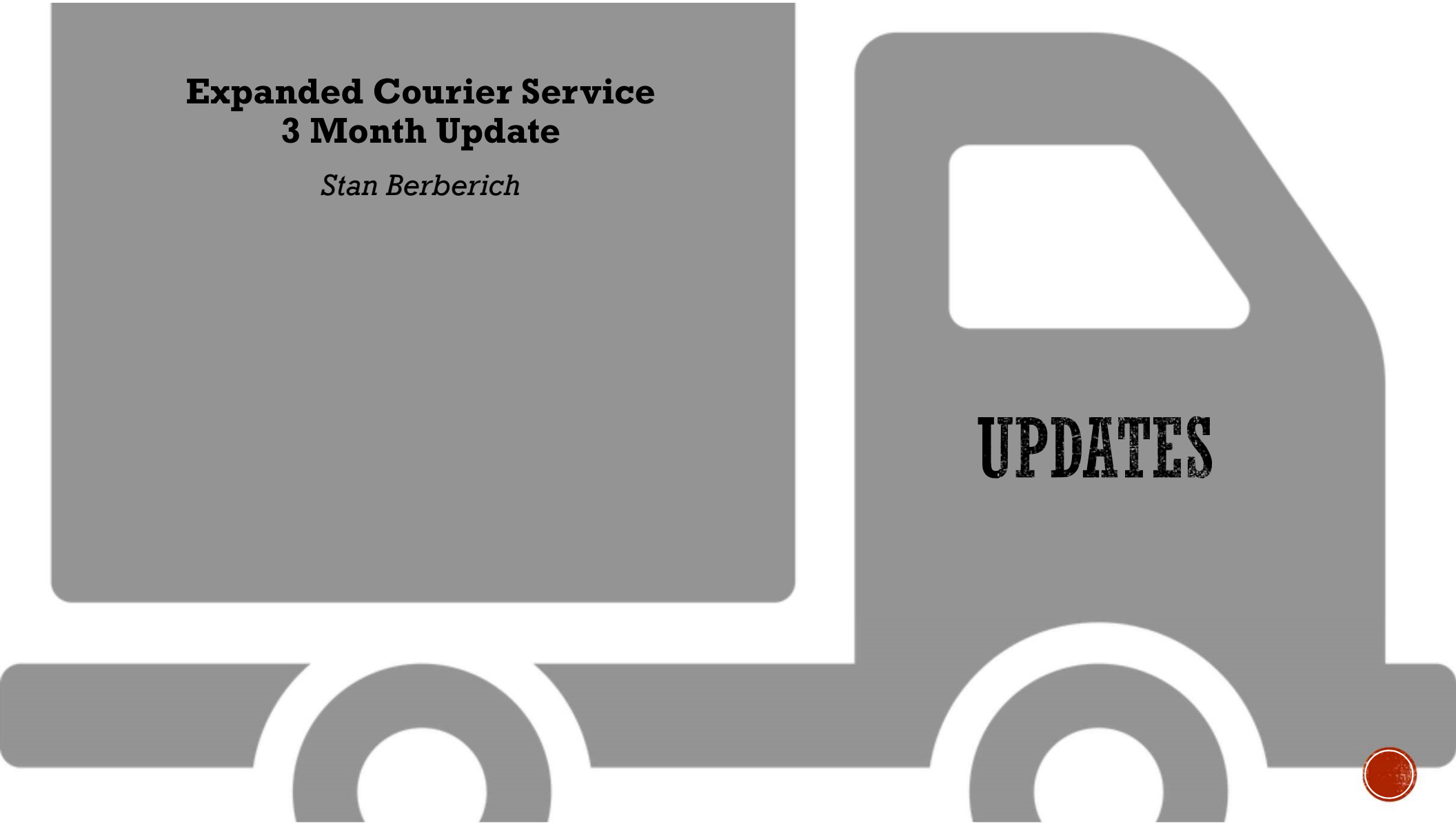
ADVISORY COMMITTEE ROLL CALL & REVIEW OF MINUTES

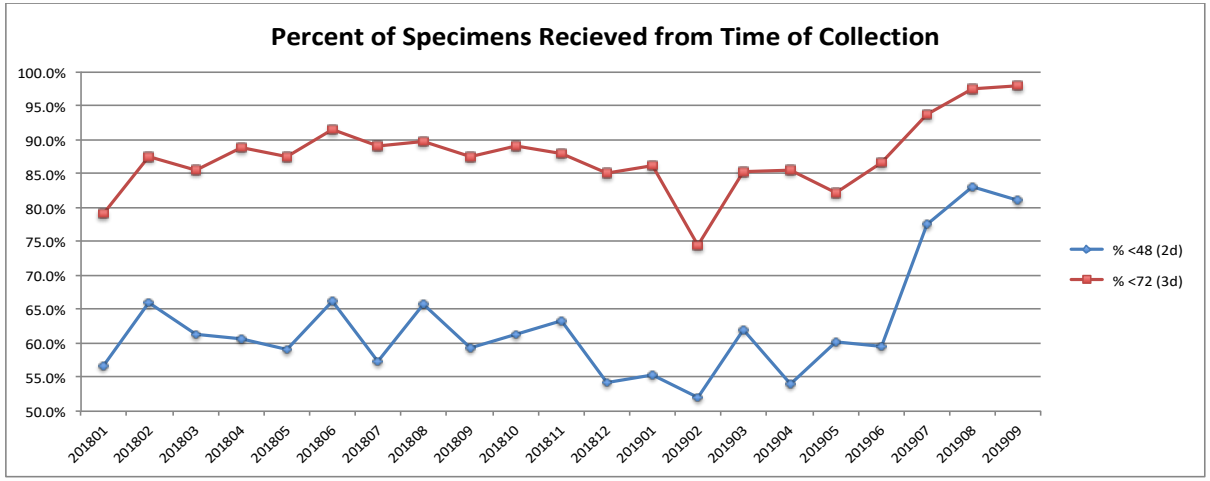
SPECIAL HEALTH SERVICES MEDIA

2019 Campaign

Expanded Courier Service
3 Month Update

Stan Berberich




Percent of Specimens Recieved from Time of Collection
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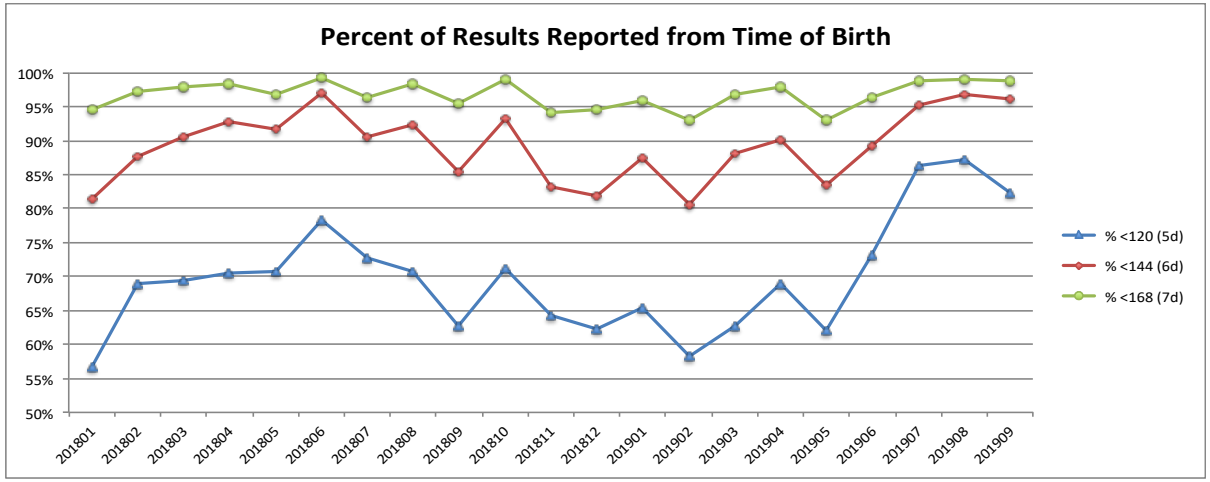
Percent of Results Reported from Time of Birth
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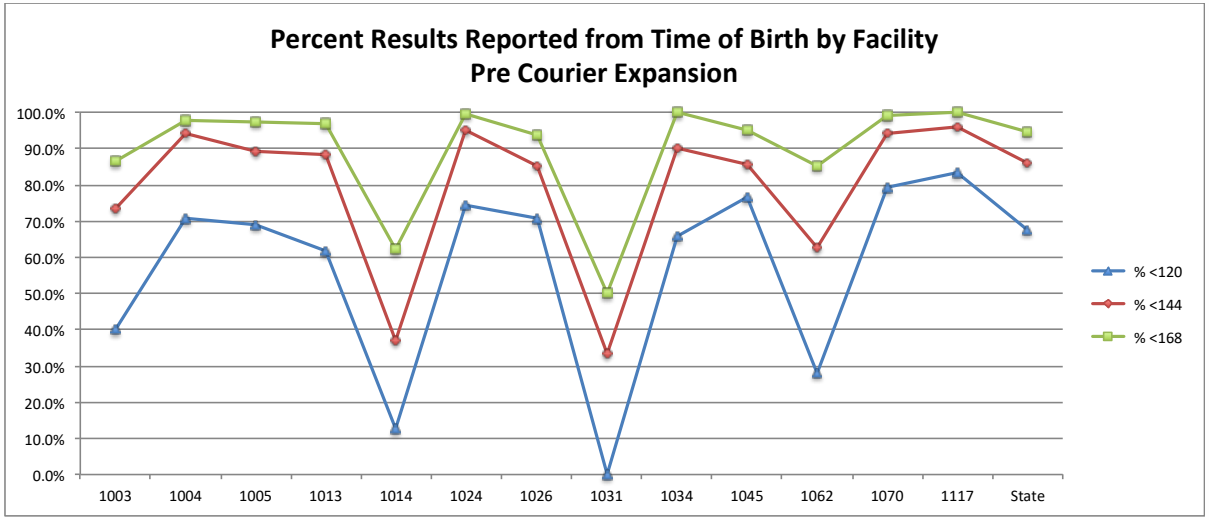
Percent Results Reported from Time of Birth by Facility
Pre Courier Expansion
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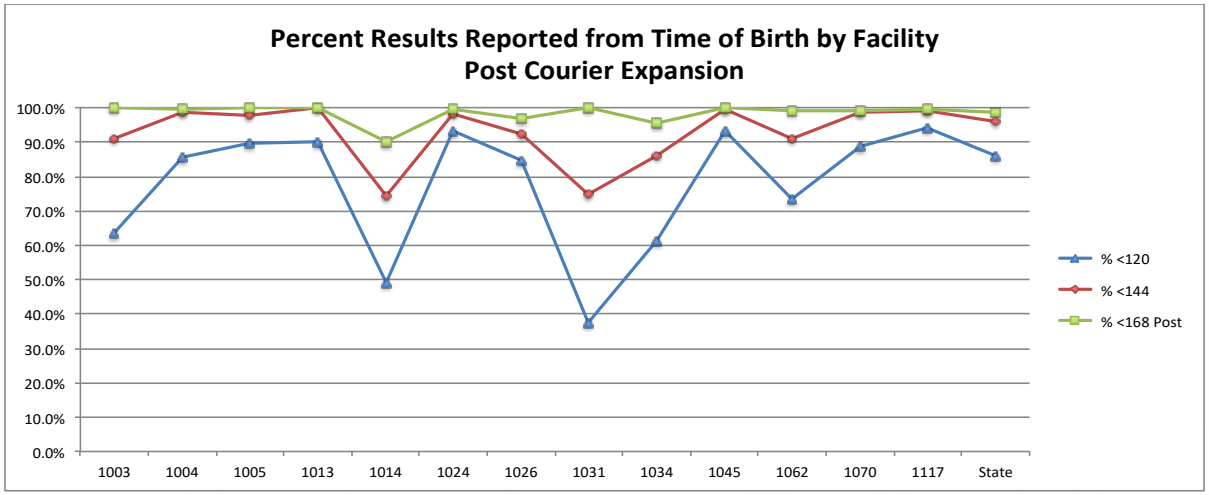
% <144
% <168


Percent Results Reported from Time of Birth by Facility
Post Courier Expansion
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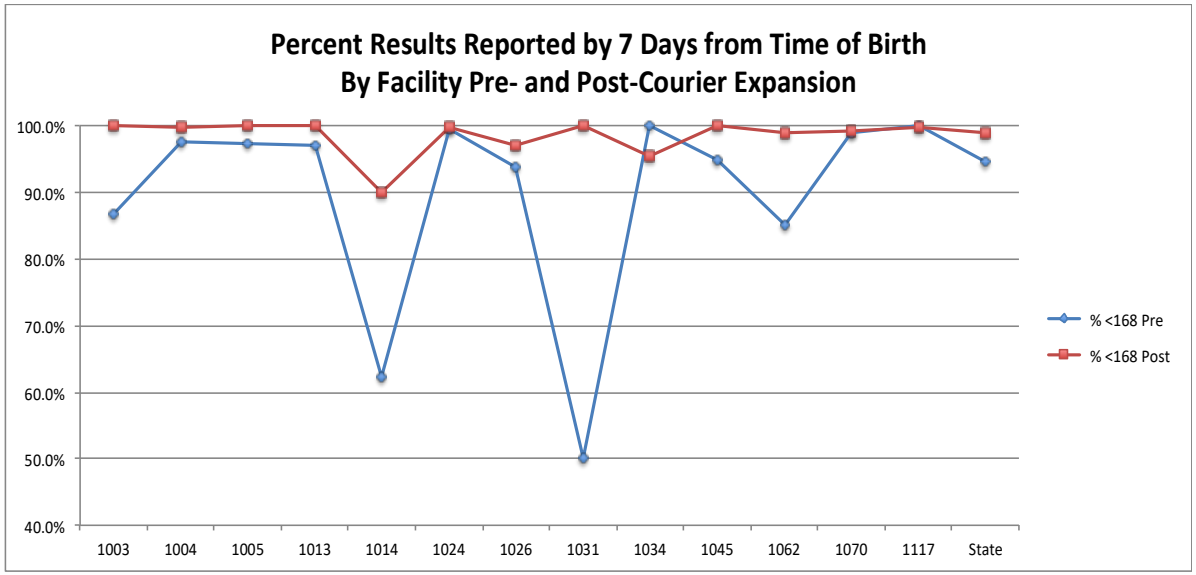
% <144
% <168 Post


Percent Results Reported by 7 Days from Time of Birth
By Facility Pre- and Post-Courier Expansion
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70.0%
60.0%
50.0%
40.0%
1003
1004
1005
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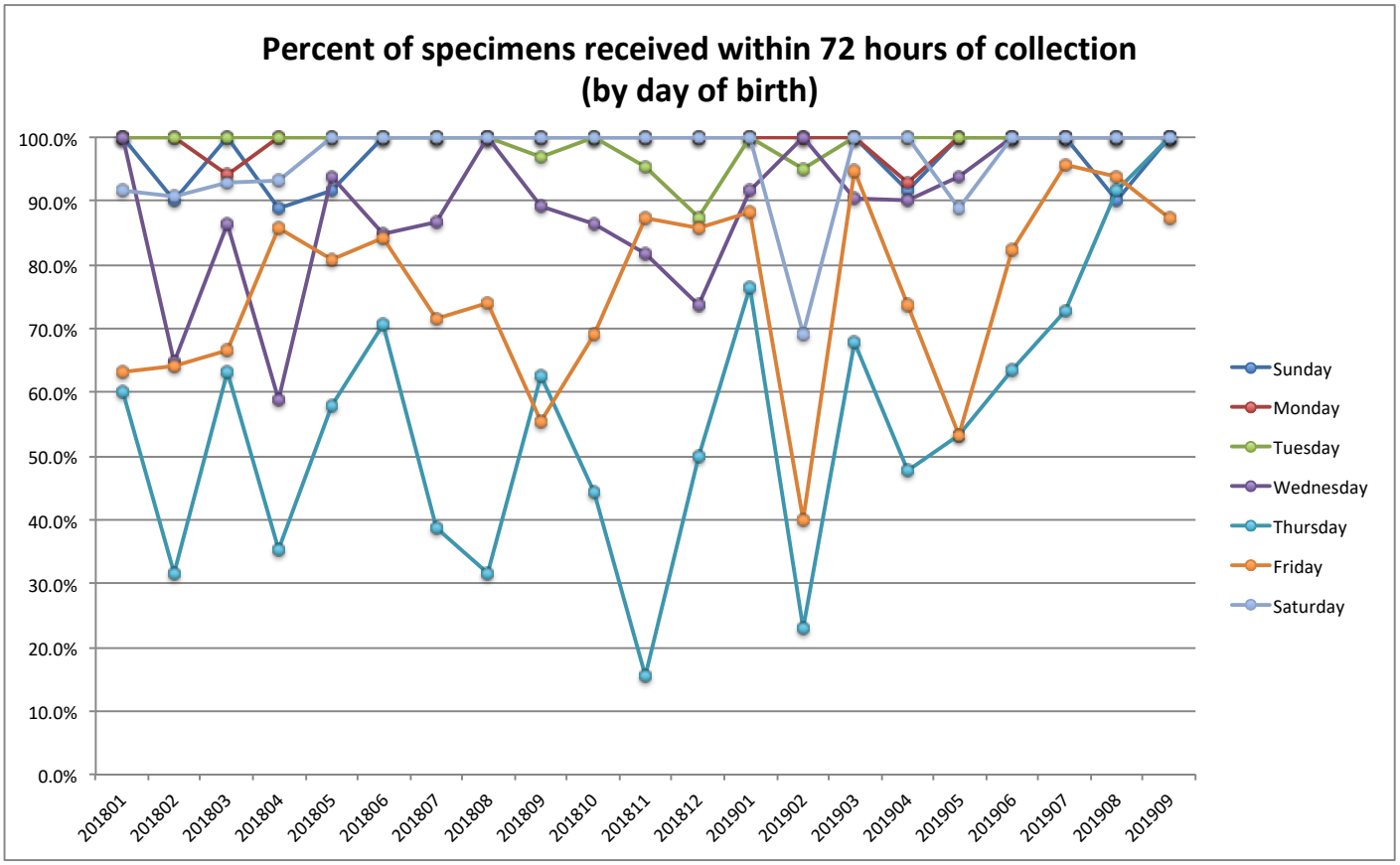
1014
1024
1026
1031
1034
1045
1062
1070
1117
State
% <168 Pre
% <168 Post


Percent of specimens received within 72 hours of collection
(by day of birth)
100.0%
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SPINAL MUSCULAR ATROPHY

Newborn Screening for Spinal Muscular Atrophy

SANFORD HEALTH

Kari Casas, MD

Sanford Health Fargo

October 1, 2019

Spinal Muscular Atrophy

- Spinal muscular atrophy (SMA): lower motor neuron disease inherited in an autosomal recessive fashion
- Clinical signs: skeletal muscle wasting and weakness (especially truncal and proximal extremity muscles), progressive respiratory compromise
- *SMN1* gene encodes SMN protein, part of the SMN complex of proteins necessary for motor neuron survival
- *SMN2* gene encodes different isoforms of SMN protein, only isoform d SMN protein is full size and functional
- ~90% of functional SMN protein from *SMN1*
- ~10% of functional SMN protein from *SMN2*

Spinal Muscular Atrophy

- Typical copy number of *SMN1*: 2
- Typical copy number of *SMN2*: 0 to 5, rarely up to 8
- ~95% of affected individuals have a homozygous deletion/gene conversion of exon 7 of *SMN1*
- Carrier rate in U.S: 1/54
- Incidence of SMA: ~one in 10,000 live births
- Types 0-IV
  - Severily depends on number of copies of *SMN2*

Spinal Muscular Atrophy

Types of SMA – Focus of Review

Type *(Alt Names)*	Age of Onset	Clinical Features	Life Expectancy	Affected Gene(s)
SMA Type 0 *(Congenital, Prenatal SMA)*	Prenatal (30-36 weeks)	Decreased fetal movements in utero, difficulty swallowing	Rarely past 6 months	**SMN1**
SMA Type I *(Severe infantile acute; Werdnig-Hoffman disease)*	Birth to six months	Cannot sit independently, difficulty breathing	24 months (med)	**SMN1** (2 SMN2 copies)
SMA Type II *(Infantile chronic)*	Six to twelve months	Sit independently, but cannot stand or walk	25 years (70%)	**SMN1** (3-4 SMN2 copies)
SMA Type III *(Juvenile, Kugelberg-Welander disease)*	After 18 months	Can stand or walk, but walking, stair-climbing become difficult. Wheelchair assistance usually needed in later life.	Normal	**SMN1** (3-4 SMN2 copies)
SMA Type IV (Adult-onset)	20-30 years	Mild to moderate muscle weakness, tremor, twitching in proximal muscles; difficulty breathing	Normal	**SMN1** (4-8 SMN2 copies)

Treatments for Spinal Muscular Atrophy

- **Spinraza (nusinersen) antisense oligonucleotide**
  - FDA approved December 2016 to treat spinal muscular atrophy in pediatric and adult patients
  - targets *SMN2*, increases production of functional SMN protein
  - administered intrathecally
  - lifelong treatment (?)
- **Zolgensma (onasemnogene abeparvovec) gene replacement**
  - FDA approved May 2019 for treatment of pediatric patients less than 2 years of age with spinal muscular atrophy
  - copy of *SMN1* in a genetically-engineered adeno-associated virus 9(AAV9) capsid
  - administered intravenously
  - one time dosage (?)

Newborn Screening for Spinal Muscular Atrophy

- From Dr. Seth Perlman's 7/20/18 presentation to the Iowa CIDAC (Congenital and Inherited Disorders Advisory Committee)
  - CDC-developed combination SCID/SMA assay
  - Targets only exon 7 deletions- no carrier detection
  - Approximately 5% false negative rate
  - Low false positive rate (0% in published NBS trials)
  - Does not detect *SMN2* copy number
  - 2nd tier *SMN2* testing would not be cost effective and would delay results by several days (similar to CF screening)

Confirmation of Diagnosis of Spinal Muscular Atrophy

- Need to confirm biallelic pathogenic variants of *SMN1*
- Depending on treatment plan, may also need to confirm SMN2 copy number and AAV9 antibody presence

Confirmation of Diagnosis of Spinal Muscular Atrophy

- Role of sponsored testing programs

Invitae SMA Panel

Confirms previous clinical diagnosis or new genetic diagnosis utilizing a more comprehensive analysis that includes read-through variants or other single base changes in the full gene. The panel provides *SMN1* deletion and *SMN2* copy number in 10 to 21 calendar days.

Invitae SMA Carrier Screen

Determines an individual's carrier status for SMA in 10 to 21 calendar days.

Invitae SMA STAT Panel

Expedited turnaround time that determines SMN1 deletion and SMN2 copy number with results provided within 4 days from sample accessioning.*

*Individual reporting times may vary

athena diagnostics

avexis

200 Forest Street, 2nd Floor • Marlborough, MA 01752 • AthenaDiagnostics.com

Athena Diagnostics operating as Quest Diagnostics

AveXis Laboratory Testing Program Enrollment

By participating in this AveXis Laboratory Testing Program, I hereby consent to the receipt by AveXis of all the data specific to the program from the laboratory service provider, Athena Diagnostics, including, but not limited to, the name, state, and National Provider Identifier (NPI) of the referring provider, test result ranges, test result values, and applicable dates. This consent is an overall consent for all testing in connection with the AveXis Laboratory Testing Program and is not given on a per order basis. I understand that costs related to the AveXis Laboratory Testing Program which includes the AveXis-Athena Test Kits and SMA Diagnostic and Antibody Tests will be billed directly to AveXis.

C.T.L. Cellular Technology Ltd.

avexis

Cellular Technology Limited (CTL) • 20521 Chagrin Boulevard • Shaker Heights, OH 44122-5350
Ph: 216-791-5084, ext 128 • Fax: 216-791-8814

AveXis Laboratory Testing Program Enrollment

By participating in this AveXis Laboratory Testing Program, I hereby consent to the receipt by AveXis of all the data specific to the program from the laboratory service provider, Cellular Technology Limited (CTL), including but not limited to, the name, state, and National Provider Identifier (NPI) of the referring provider, test result ranges, test result values, and applicable dates. This consent is an overall consent for all testing in connection with the AveXis Laboratory Testing Program and is not given on a per order basis. I understand that costs related to the AveXis Laboratory Testing Program which includes the AveXis-CTL Test Kits and ELISA: AAV9 Antibody Tests will be billed directly to AveXis.

SPINAL MUSCULAR ATROPHY

JOINING IOWA PILOT

Newborn Screening Laboratory Assay for SMA

Travis Henry, PhD

Iowa Newborn Screening Laboratory

State Hygienic Laboratory at the University of Iowa

ACHDNC and the RUSP

- “Expand the Recommended Uniform Screening Panel (RUSP) to include the addition of spinal muscular atrophy (SMA) due to homozygous deletion of exon 7 in SMN1”
- Letter to Secretary of Health and Human Services Alex Azar from Joseph A. Bocchini, Jr., M.D., ACHDNC Chairperson

SMA Pathogenesis

Genetics:

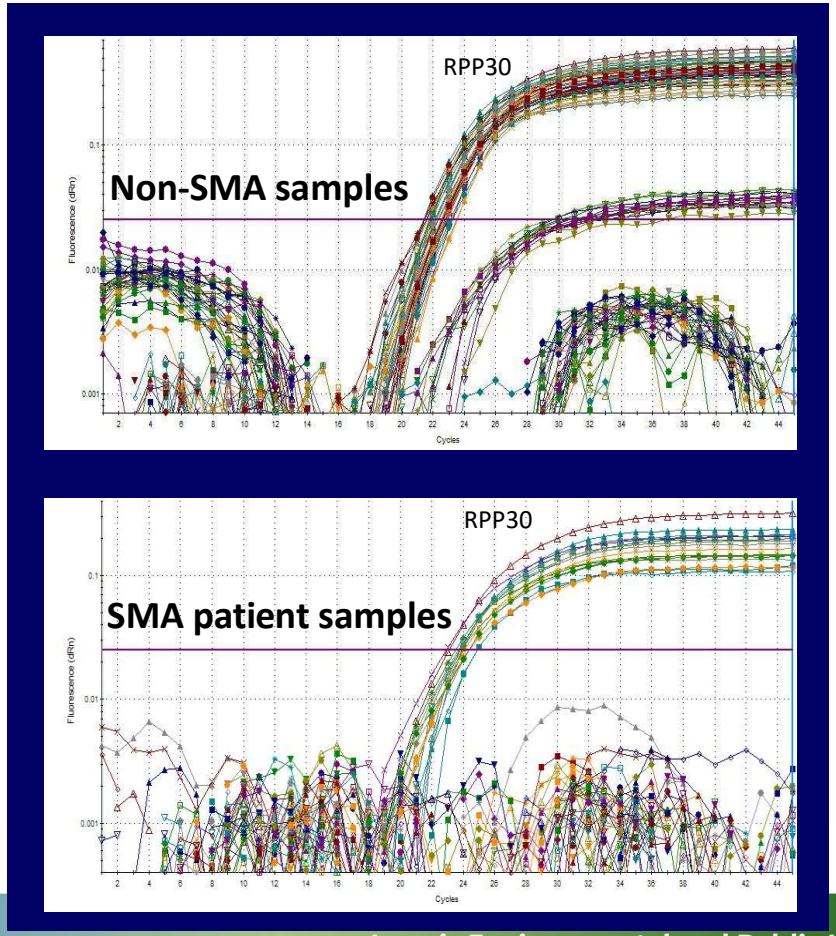
- 5q-SMA Types I-IV
- ~95% - Homozygous deletion/gene conversion mutation of Survival Motor Neuron 1 (SMN1) exon 7
- ~5% - compound heterozygotes
- Variable number of SMN2 copies, correlates with phenotype

Screening:

- Screening Target: homozygous deletion of SMN1 exon 7
- Method: Quantitative Real-time Polymerase Chain Reaction (qPCR) assay using dried-blood spots (same as SCID)
- Additional/optional testing: SMN2 dosage (informs phenotype)

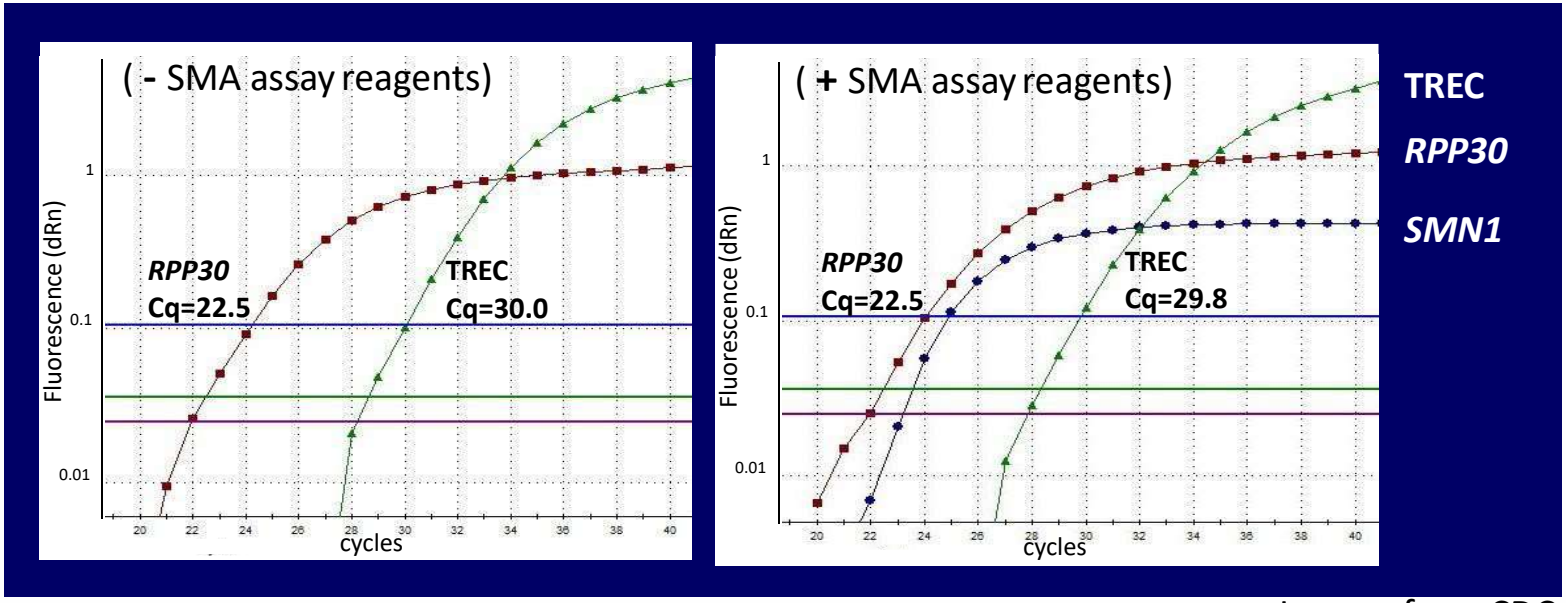
Assay

- SMA assay targets homozygous deletion of SMN1 exon 7: 95% of cases
- SMA assay is a binary assay due to homozygous deletion: SMN1 is either present or absent



Images from CDC

The SMA assay can also be multiplexed with SCID



Images from CDC

- Cq values for RNase P and TREC are unaffected by the addition of reagents for SMA

Assay Changes

- SCID assay is currently an *in situ* assay: 2.0mm punch is added to a PCR plate, cleaned, and then PCR reagents added to cleaned punch for amplification of TREC
- SMA assay will be a separate DNA extraction using a 3.2mm spot and DNA and PCR reagents added to a 384 well PCR plate
- Advantages:
  - Improved specimen tracking
  - Improved workflow (4 x 96 well plates = 384 well)

Timeline

- Now until March 2020: develop lab assay and define reference ranges
- March 2020 to July 2020: QA exercise with blinded specimens
- July 2020: begin SMA pilot
- July 2021: proposal to IDPH/SBOH to add SMA to the Iowa newborn screening panel (pending successful pilot)

Questions?

- Travis Henry PhD
  - travis-henry@uiowa.edu
  - (319) 335-4364

NICU Newborn Screening Collection: "To Hold or Not To Hold: That Is The TPN Question" Sanford NICU experience

Mohamed Mohamed, MD
Syrina Abarqoub, MD

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Introduction

- Premature infants are known to have a higher rate of false positive results (FP)
  - Illness related stress
  - Use of screening cutoffs based on studies of term infants
  - Use of total parenteral nutrition (TPN)
  - Liver immaturity

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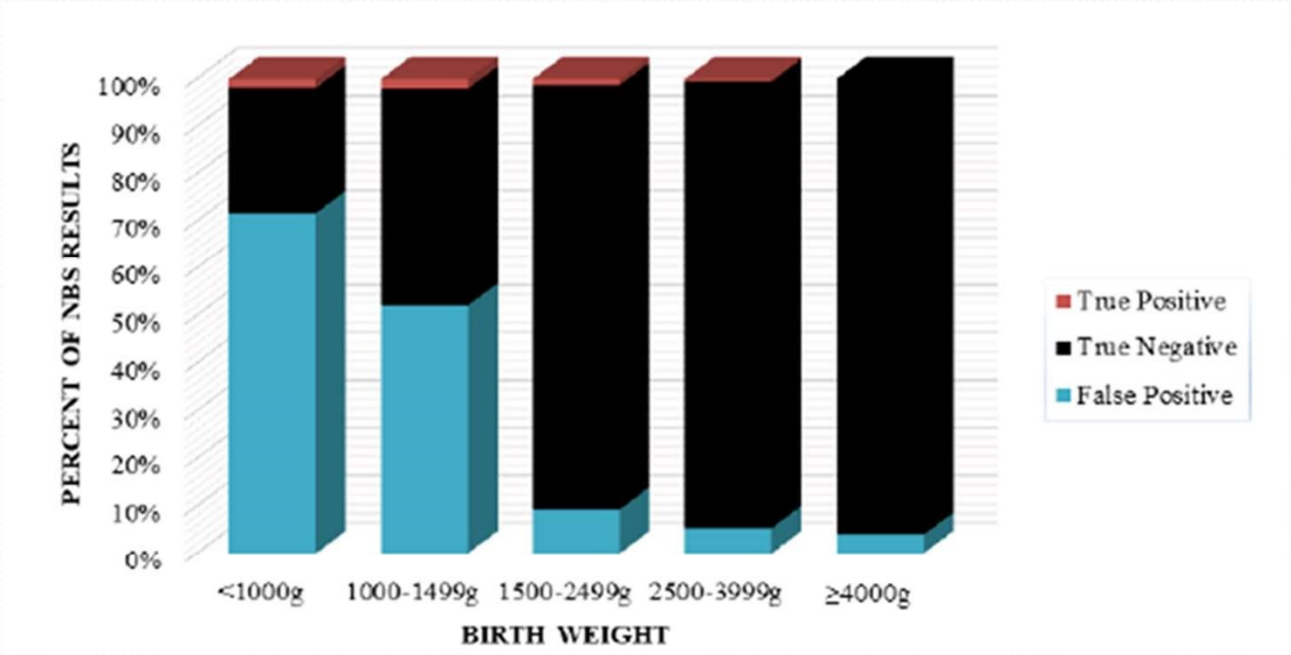

Asghar A et al J of Child and Adolescent Health (2019)

Why to care about FP results

Effect of FP results:

- Unnecessary additional blood testing
- Parental anxiety, stress and long-term worry
- May affect parent-child relationship
- Additional health care cost

1.Waisbren SE, et al. *JAMA*. 2003.
2. Beth A. Et al, *Pediatrics* 2012

Neonatologist reaction in recent years

It is unavoidable human nature that too many false alarms can make us less vigilant in our attention to the valid ones.

How to reduce FP?

- Retrospective cohort study included 539 NICU admissions.
- TPN was replaced with 10% D10W for 3h before sample collection.
- The new protocol reduced false-positive results for each birth weight group by at least 50% and overall by 74% (P = 0.008)
- The protocol reduced estimated costs by >80%.

Morris M, et al, Genet Med. 2014

Reduction in newborn screening metabolic false-positive results following a new collection protocol

	Preintervention (N = 274)		Postintervention (N = 265)	
	Negative, n (%)	False-positive, n (%)	Negative, n (%)	False-positive, n (%)
Birth weight				
<1,000 g	13 (65.0)	7 (35.0)	14 (82.4)	3 (17.6)[a]
1,001–1,500 g	23 (92.0)	2 (8.00)	24 (96.0)	1 (4.00)[b]
>1,501 g	222 (96.9)	7 (3.06)	223 (100)	0 (0.00)[c]
Total	258 (94.2)	16 (5.83)	261 (98.5)	4 (1.50)[d]

CI, confidence interval; OR, odds ratio.

[a] x_1^2 = 1.4; P = 0.29 (Fisher's exact test); OR = 1.98; 95% CI: 0.61–6.5. [b] x_1^2 = 0.4; P = 1.0 (Fisher's exact test); OR = 2; 95% CI: 0.19–20.7. [c] x_1^2 = 6.9; P = 0.02 (Fisher's exact test). [d] x_1^2 = 7.1; P = 0.008; OR = 3.87; 95% CI: 1.31–11.42.

Morris M, et al, Genet Med. 2014

- Holding TPN containing AAs for 3 hours before NBS collection is a practical and cost-effective method to significantly reduce the false-positive rate for AA in VLBW infants (3.1% vs 11.8%; P = .037). Tim At et al. J pediatr. 2015

- Withhold total parenteral nutrition 4 hours prior to obtaining NBS sample did not significantly reduce FP, however, there was significant reduction in FP results in abnormalities of AC profile in patients <1000 g. Asghar A et al, J of Child and Adolescent (2019

Quality Improvement Project

- Aim: to reduce NBS-FP results in the NICU caused by elevated aa or acylcarnitine abnormalities
- A multidisciplinary quality improvement team reviewed the literature and developed a new newborn screen collection protocol
- A comprehensive nursing/medical staff educational initiative was conducted to improve techniques for obtaining samples and to provide an overview of the changes to the NBS guideline
- Period 1: Implemented in July 2014
- Period 2: Implemented in Sept 2015

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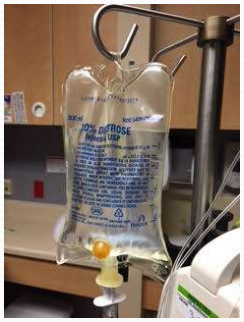
Intervention- period 1

- **July 2013 –June 2014**
- NBS specimen collected regardless of TPN administration
- Sample can be obtained from central line
- Carnitine added to TPN day 1

- **July 2014 –New Protocol**
- Stop TPN for 4h prior to collection of NBS
- Infuse D10%w while TPN stopped
- Always heal stick sample
- No carnitine for 7 days



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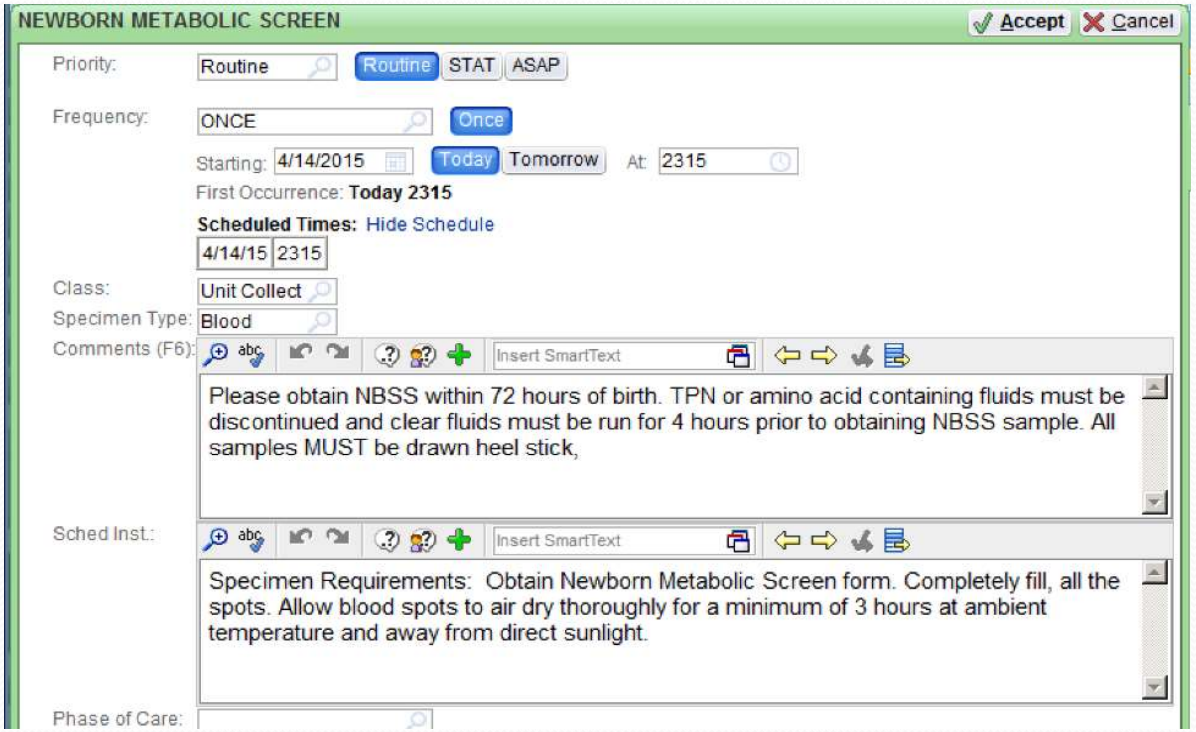
Implementation- period 1

- Teaching for providers and nurses
- Clear instructions added to admission orders

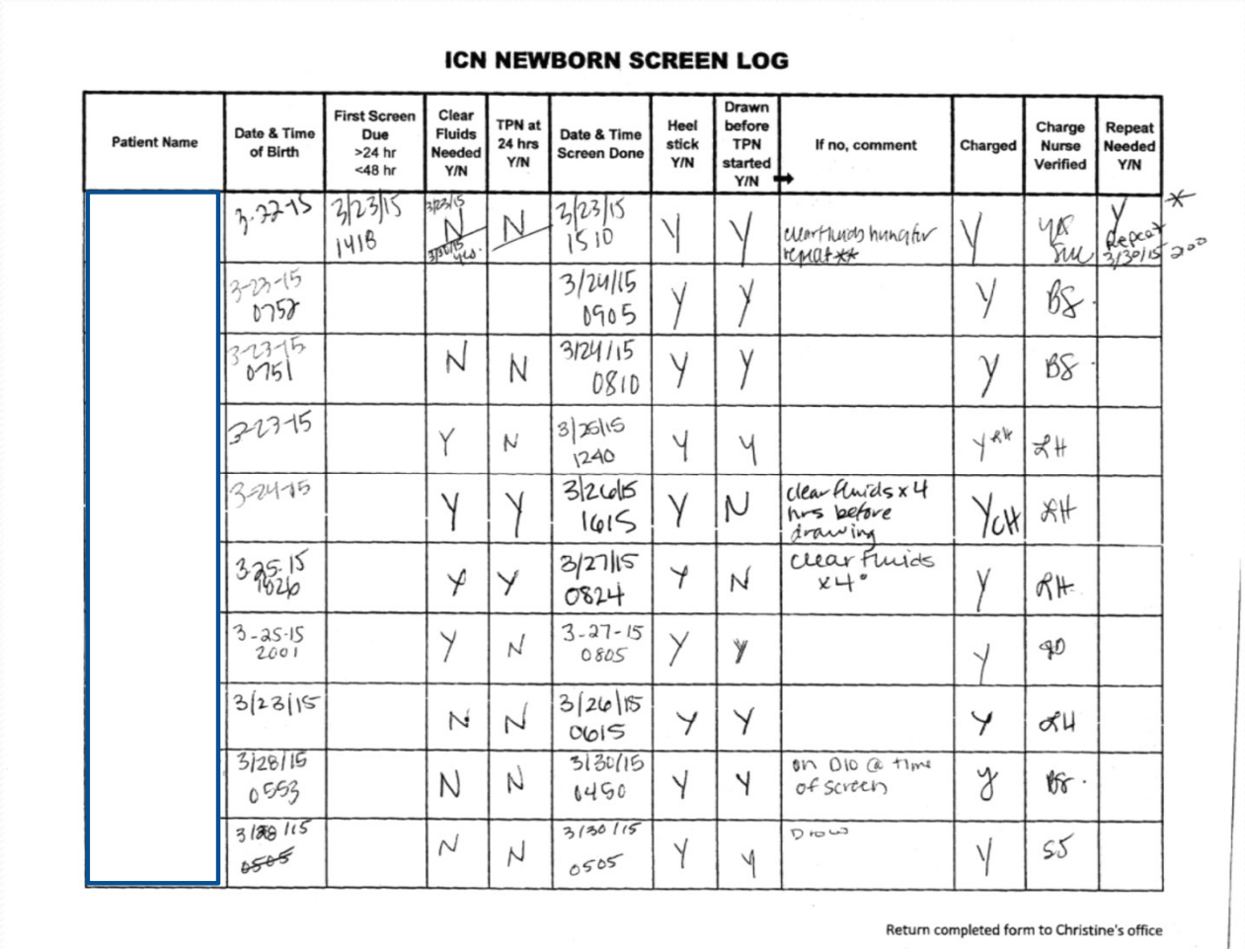


Intervention- period 2

- The NBS collection protocol modified the length of TPN interruption for infants with birth weight less than 1000 grams to 6 hours instead of 4 hours.

- All other intervention remained the same.

Data tracking

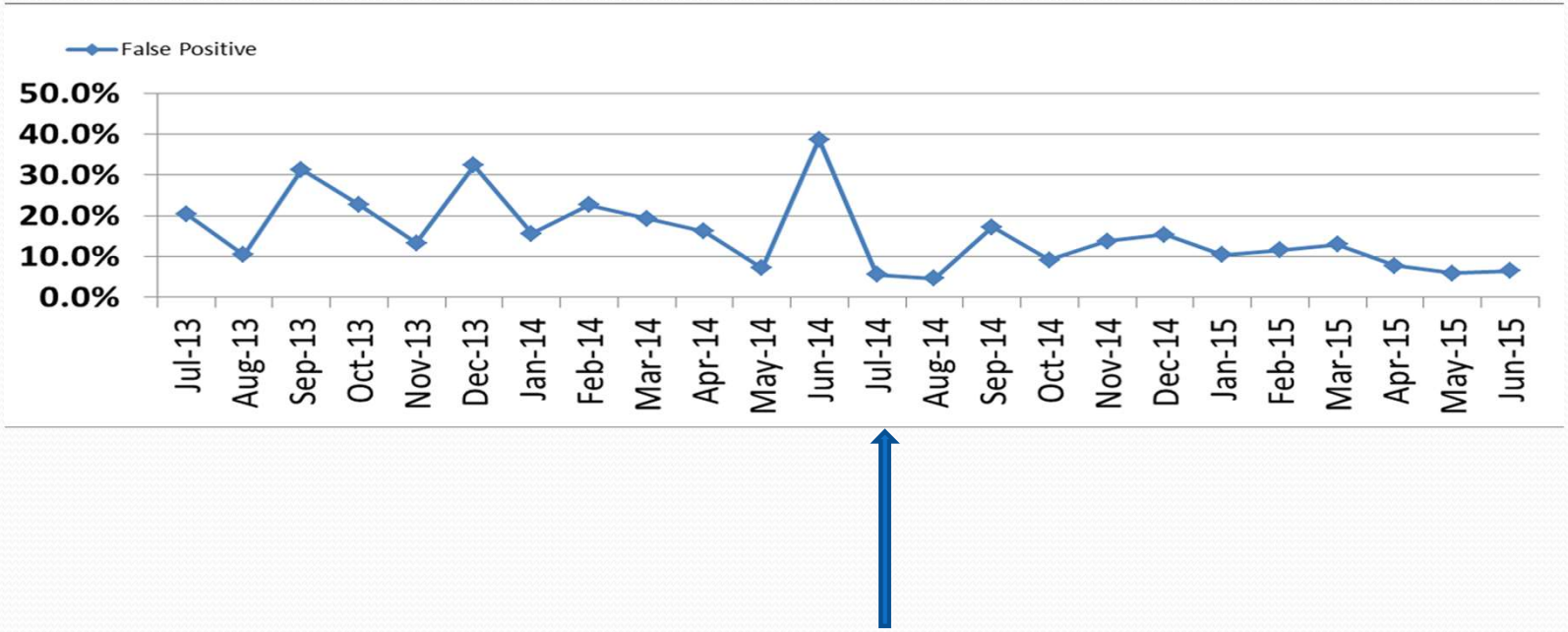


ICN NEWBORN SCREEN LOG

Patient Name	Date & Time of Birth	First Screen Due >24 hr <48 hr	Clear Fluids Needed Y/N	TPN at 24 hrs Y/N	Date & Time Screen Done	Heel stick Y/N	Drawn before TPN started Y/N	If no, comment	Charged	Charge Nurse Verified	Repeat Needed Y/N
	3.22.15	3/23/15 1418	3/23/15 N 3/30/15 yes	N	3/23/15 1510	Y	Y	overflowing hung for repeat**	Y	YR SW	Y * Repeat 3/30/15 2nd
	3-23-15 0758				3/24/15 0905	Y	Y		Y	BS	
	3-23-15 0751		N	N	3/24/15 0810	Y	Y		Y	BS	
	3-23-15		Y	N	3/25/15 1240	Y	Y		Y RK	RH	
	3-24-15		Y	Y	3/26/15 1615	Y	N	clear fluids x 4 hrs before drawing	Y CH	RH	
	3.25.15 1020		Y	Y	3/27/15 0824	Y	N	clear fluids x 4°	Y	RH	
	3-25-15 2001		Y	N	3-27-15 0805	Y	Y		Y	RD	
	3/23/15		N	N	3/26/15 0615	Y	Y		Y	RH	
	3/28/15 0553		N	N	3/30/15 0450	Y	Y	on D10 @ time of screen	Y	BS	
	3/28/15 0505		N	N	3/30/15 0505	Y	Y	D10	Y	SS	

Return completed form to Christine's office

Results- period 1



Intervention
July 2014

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Results by birth weight- period 1

	Pre-intervention June,2013- July,2014		Post-intervention July,2014- June,2015		P-value
Birth Weight	Total No	FP, n(%)	Total No	FP, n(%)	
<1000	26	18(69.2)	23	11(47)	0.1548
1000-1500	37	24(64.9)	28	7(25)	0.0024
>1500 g	349	43(12.3)	351	25(7.1)	0.0217
Total	412	85(20.6)	402	43(10.7)	0.0001

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Period 2: 4 hrs Vs 6 hrs TPN interruption in preterm < 1000g

	Post-intervention July, 2014- June, 2015		6 hours TPN interruption Sept, 2015 to May, 2019		P-value
Birth Weight	Total No	FP, n(%)	Total No	FP, n(%)	
<1000	23	11(47)	111	24(22)	0.0243

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Cost analysis

Item	Pre-intervention	Post-intervention
Confirmatory testing	23,265	11,632
Supplies for testing	230	115
Supplies for new protocol	0	1200
Total	23,495	12,947

Cost analysis based on estimated 400 NICU admissions per year with 20% FP pre-intervention and 10% post-intervention.
Supplies for testing: lancet and heel warmer for blood draws, and urine bag for urine tests. New protocol supplies: bag of D10W, intravenous spike, and syringe.
Professional time of nurses, NICU providers and genetic specialist are not included in the cost analysis.

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Pediatrics
January 2018, VOLUME 141 / ISSUE 1 MeetingAbstract
Section on Neonatal-Perinatal Medicine Program

Reduction in Newborn Screening False Positive Results in the Nicu: A Quality Improvement Project

Mohamed W. Mohamed, Jo-Ann Smith, Stephen Nelson, Kari Casas, Waseem Altaf

BACKGROUND: Premature infants are known to have a higher rate of false positive newborn screening (NBS) results that can lead to unnecessary additional blood testing,

Concerns

- Holding TPN in the first few days of life for 4-6 hrs in preterm infants can affect aa intake.

- Early administration of amino acids is associated with a positive nitrogen balance but no evidence it affects mortality, early and late growth and neurodevelopment.
  Trivedi at al, Cochrane Database Systematic Rev. 2013.

- We make up the amount of protein by increasing the total amount of protein in the TPN on the day of TPN interruption.

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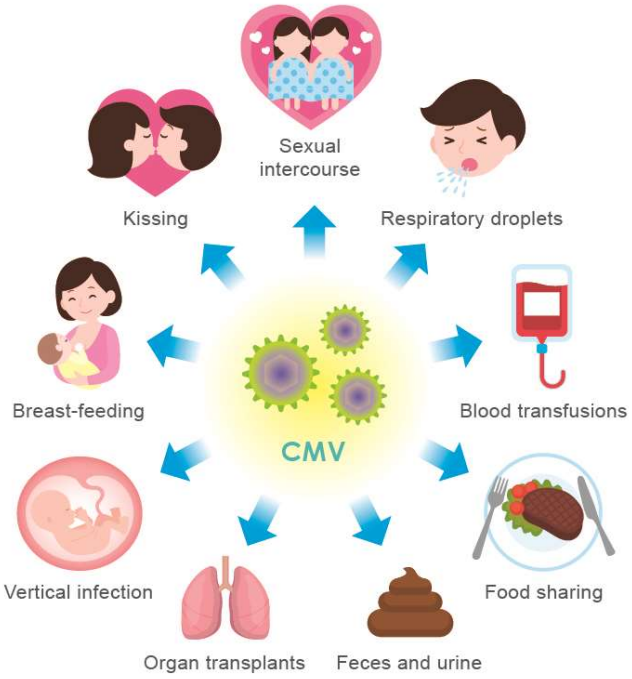
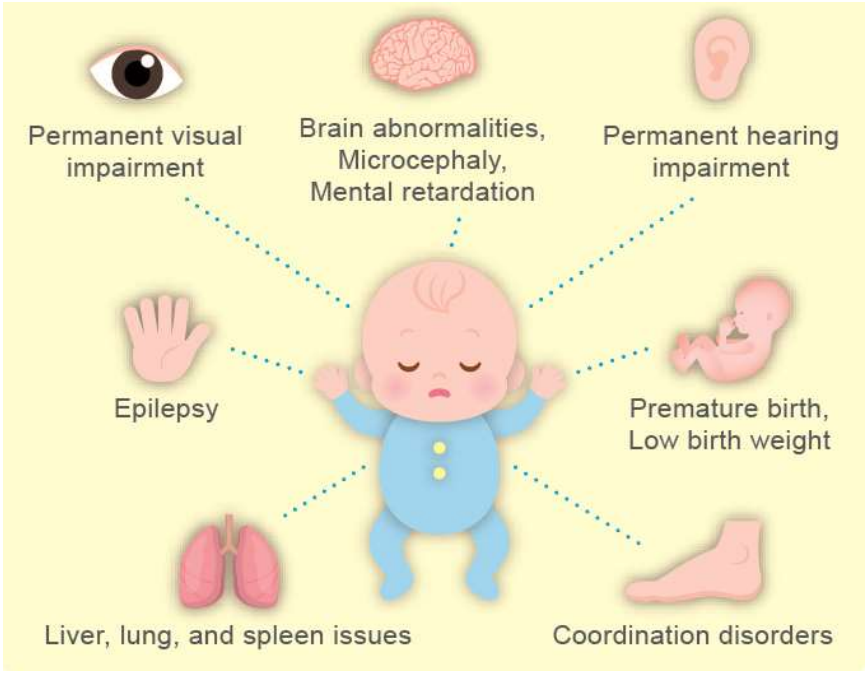
Conclusion

- New protocol easy to implement, no adverse events
- 50% reduction in FP results related to elevated aa or acylcarnitine abnormalities (20.6% vs 10.6%)
- Significant reduction in cost, number of tests, and blood loss.
- Anticipated decrease in parental anxiety
- Saves providers time and effort
- No false negatives known to date

CONGENITAL CYTOMEGALOVIRUS

Wendy Bates – Advocate and Mother to a Child With CCMV





June 2, 2018

Emilia Jo Bates


1


LITTLE
BUT
FIERCE

ACHDNC
Secretary's Advisory Committee on Heritable Disorders in Newborns and Children

COMMUNICATION OF NBS RESULTS:

A TOOL FOR PROVIDERS

Share the specific [positive]* newborn screening result and associated condition(s) with the family.
- Help the family understand that a [positive]* newborn screening result is serious, but that you are there to help guide them through the next steps.

Comprehension: Assess the family's understanding of newborn screening.
- Assess if the family recalls and understands the process of newborn screening.

Reiterate what screening is and is not.
- Remind the family about the purpose of newborn screening and that it is not a diagnostic test, so it is important that timely follow-up confirmatory testing be done.

Engage with the family and provide information at their desired level and pace.
- Offer to provide the family additional result-specific information provided by the state newborn screening program.
- Discuss information using non-medical terms, at the family's pace and desired level of detail.

Explore the family's emotions.
- Explore with the family how they might use their support system or other support resources now and as they go through the diagnostic process.
- Remember there is a wide spectrum of how families may cope with this result (anxiety to denial). Tailor your discussion to help the family hear and retain the information discussed.

Next steps: Discuss a shared plan and provide resources.
- Discuss with the family a shared plan that is concrete, specific, and includes the following:
  - Where, when, and with whom is the next appointment?
  - What testing will be considered and/or done?
  - What should they watch for in their child while they wait?
  - Who can they contact if they have additional questions or concerns?
- Assess the family's understanding of the visit and information provided using teach-back methods, and provide valid websites for them to get more information.

A positive newborn screening result can also be referred to as an abnormal result, an out-of-range result, or presumptive positive result.

For more information about the Advisory Committee on Heritable Disorders in Newborns and Children, please visit https://www.hrsa.gov/advisory-committees/heritable-disorders

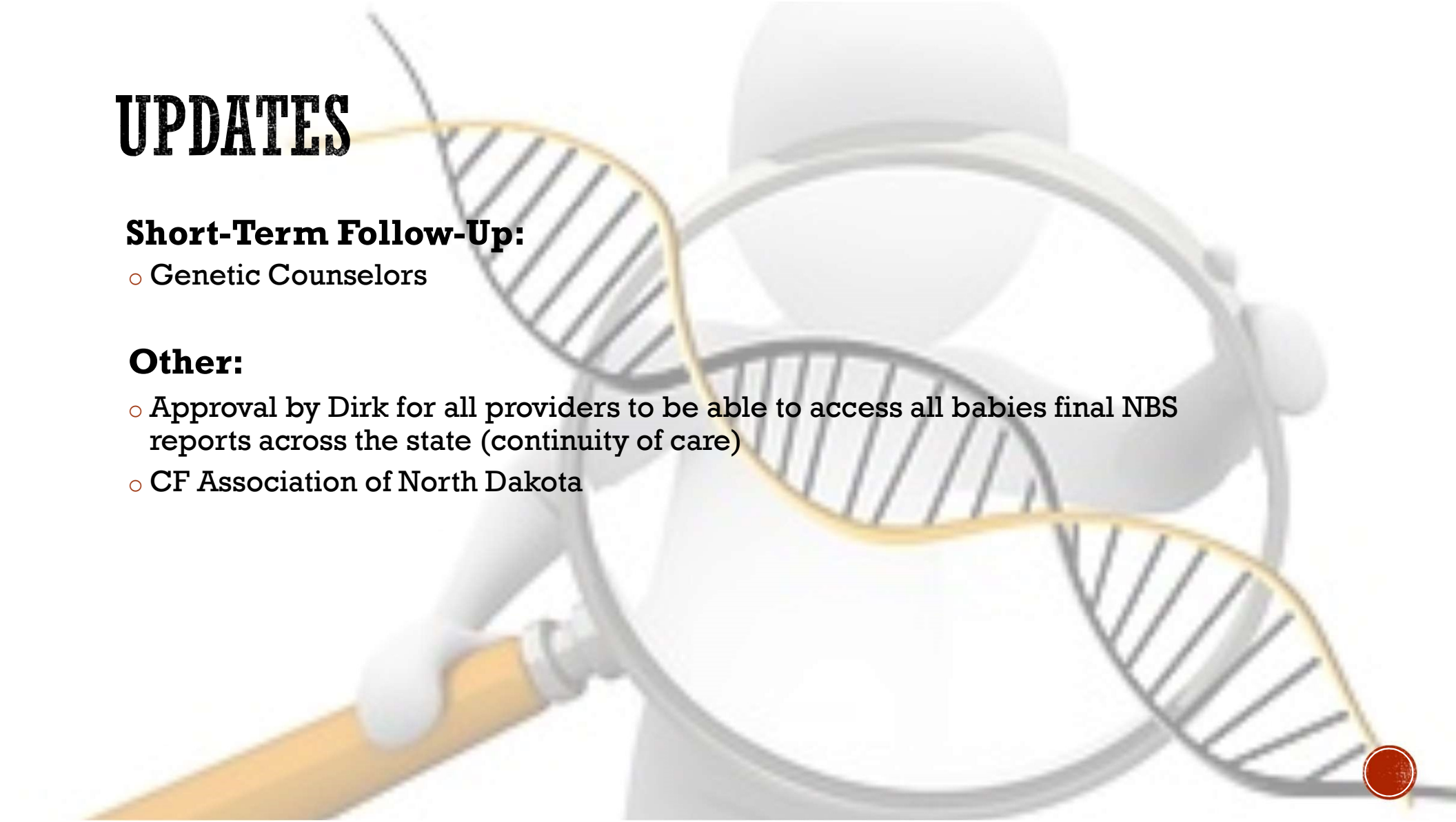
UPDATES

Short-Term Follow-Up:

- Genetic Counselors

Other:

- Approval by Dirk for all providers to be able to access all babies final NBS reports across the state (continuity of care)
- CF Association of North Dakota



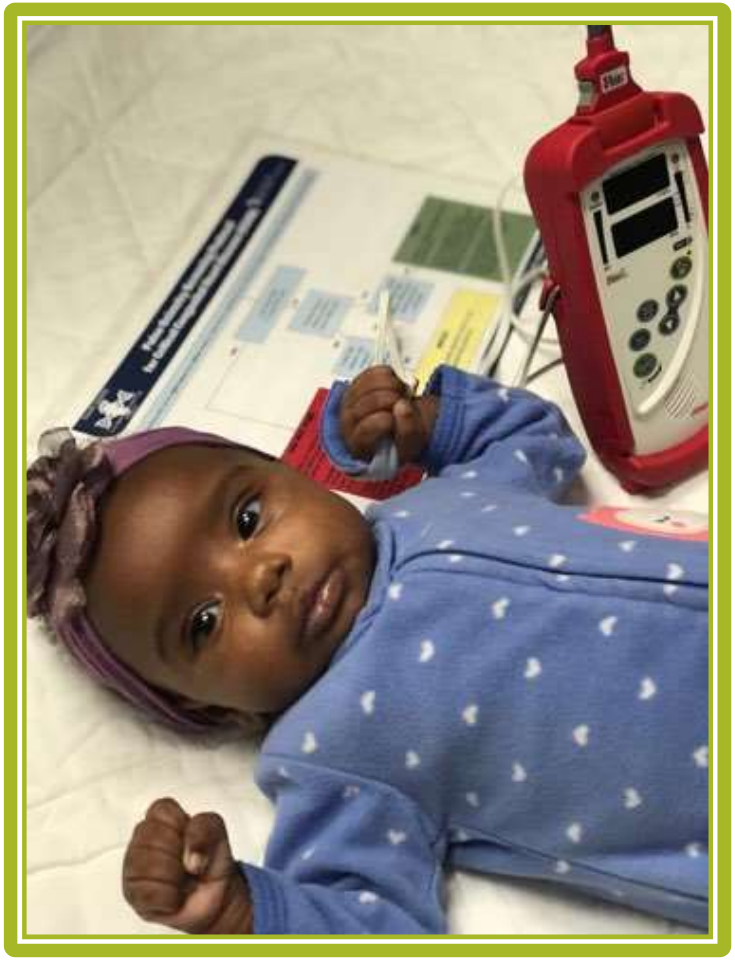
UPDATES

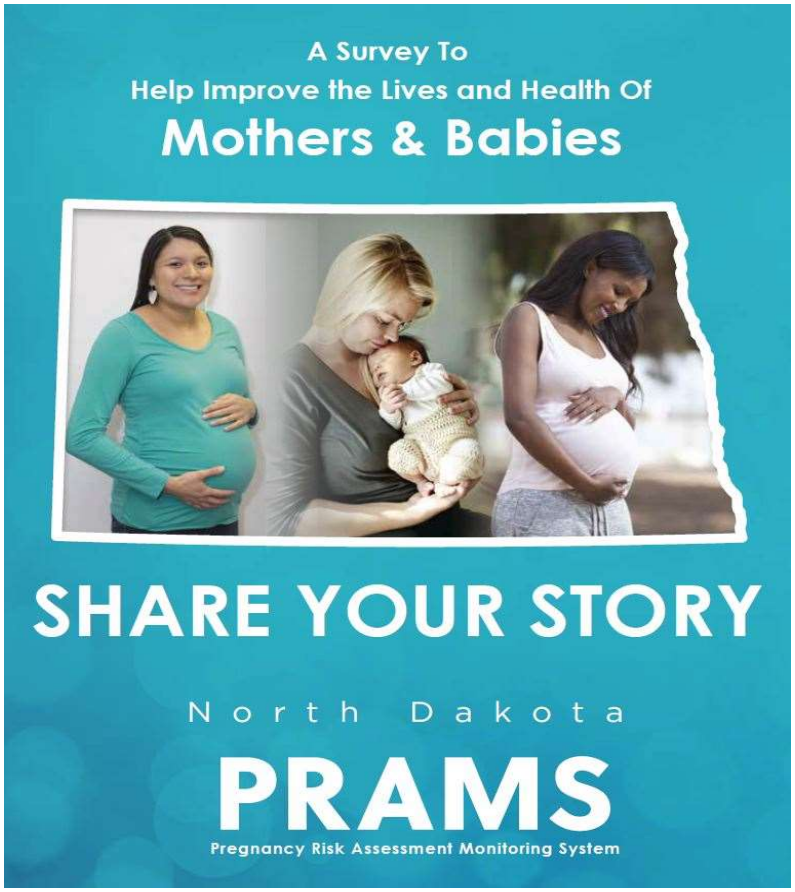
Long-Term Follow-Up:

- Training modules
- Video production
- **19** children identified with a disorder through NBS thus far
  - **5** CH
  - **4** TMS (3 carriers)
  - **3** HGB (114 carriers)
  - **3** CAH
  - **1** CF (10 carriers), **2** CRMS
  - **1** BTD
  - *1 CF (2 copies deltaF508/on tx) & 2 HGB (1 SS & 1 CC)are pending diagnosis*
- 3 children identified through NBS are MN residents:
  - SHS tri-state resource listing (MN, SD, MT)

PRAMS Data

Presenter: *Grace Njau*
ND DoH Epidemiologist





North Dakota PRAMS

Tribal Nations TNRG Research Group

NDSU NORTH DAKOTA STATE UNIVERSITY

NORTH DAKOTA PRAMS

Grace Njau, Director

RUTGERS
Edward J. Bloustein School of Planning and Public Policy

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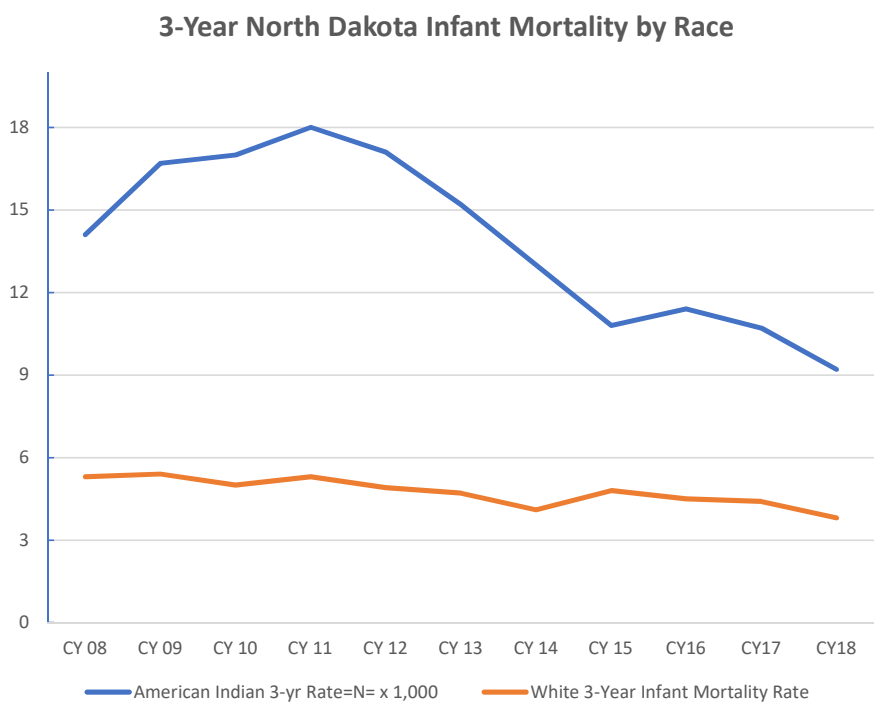
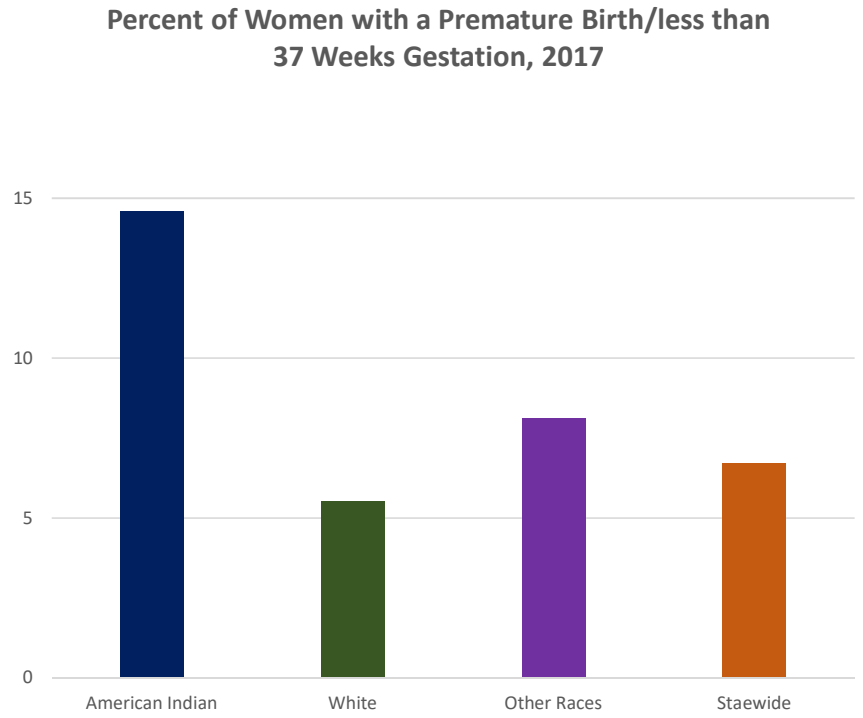
ND PRAMS AIMS

- ND PRAMS Specific Aims:
  - Collecting, analyzing, and disseminating data aimed at reducing infant and maternal morbidity and mortality
  - Fostering culturally competent research practices with tribal populations in ND
- *Long-term Goal*: To reduce maternal and infant morbidity and mortality in North Dakota through surveillance, partnerships, and resource allocation

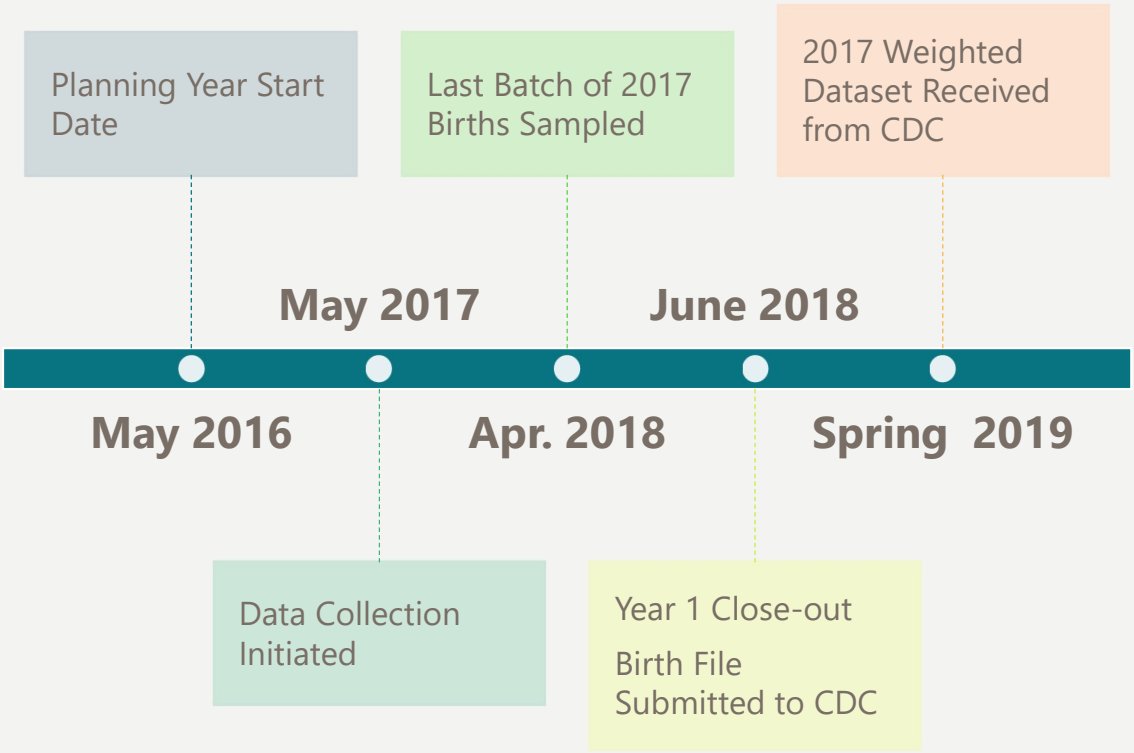
NORTH DAKOTA BIRTH OUTCOMES



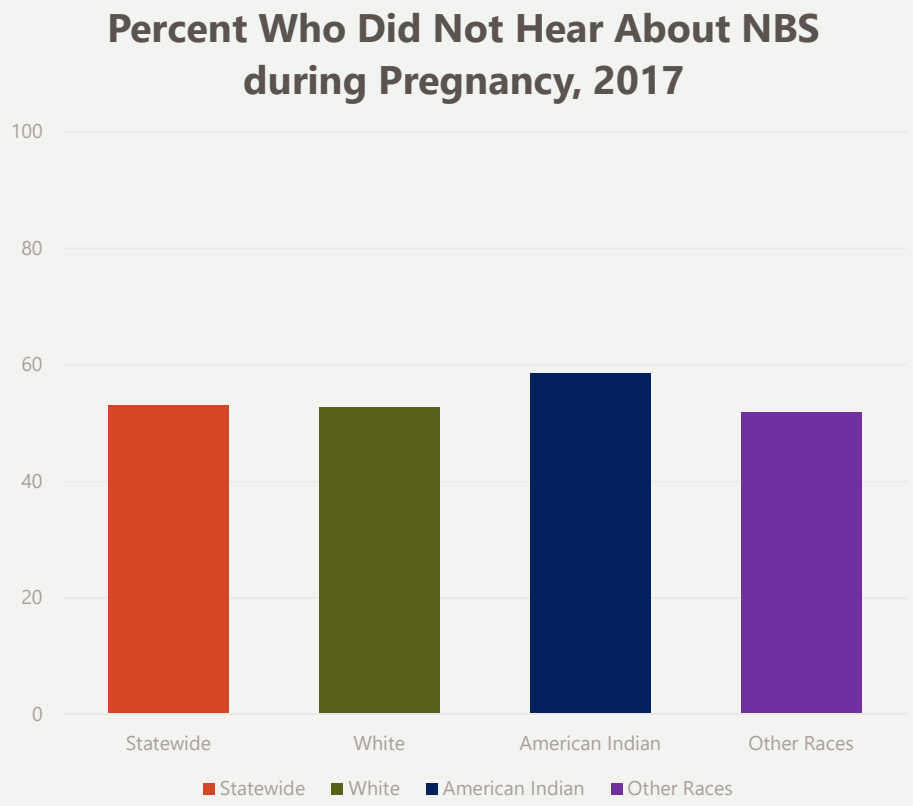


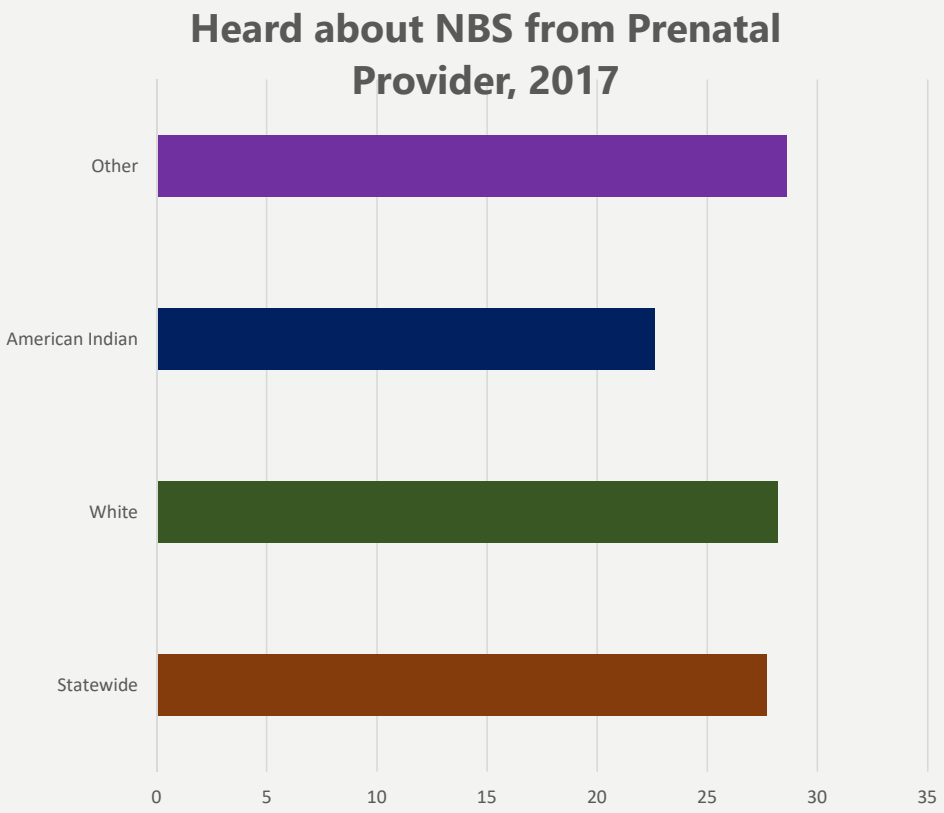
- 7% of all births were premature
- American Indian infant more likely to be born premature than non AI at 14%
- 4 per 1,000 White infant deaths vs. 9 per 1,000 American Indian infant deaths

ND PRAMS PROJECT TIMELINE

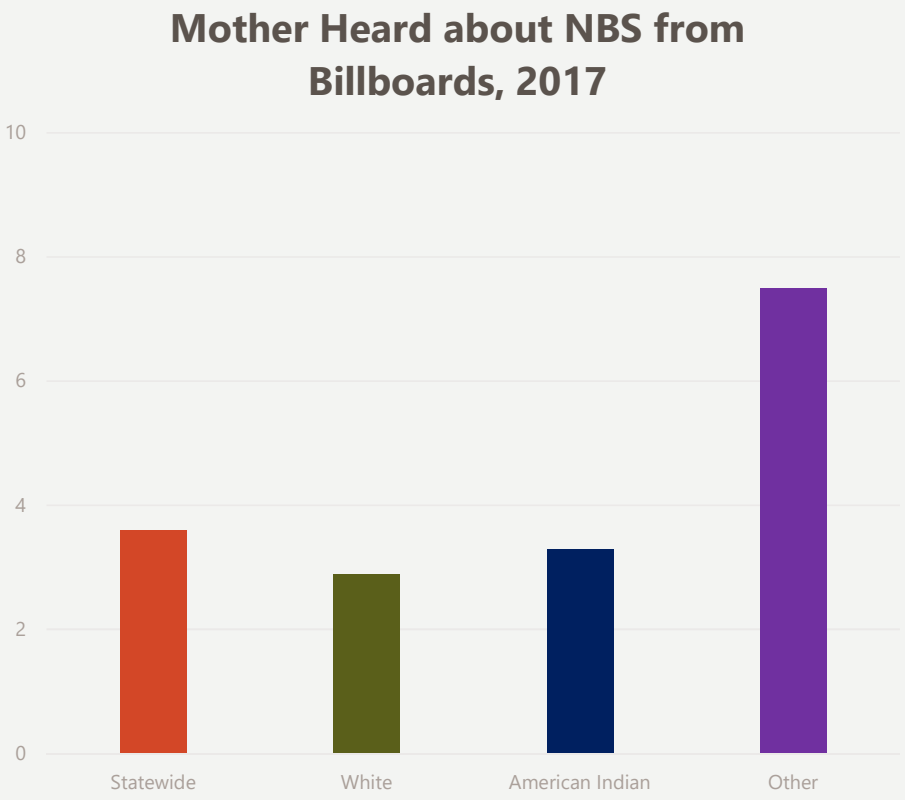


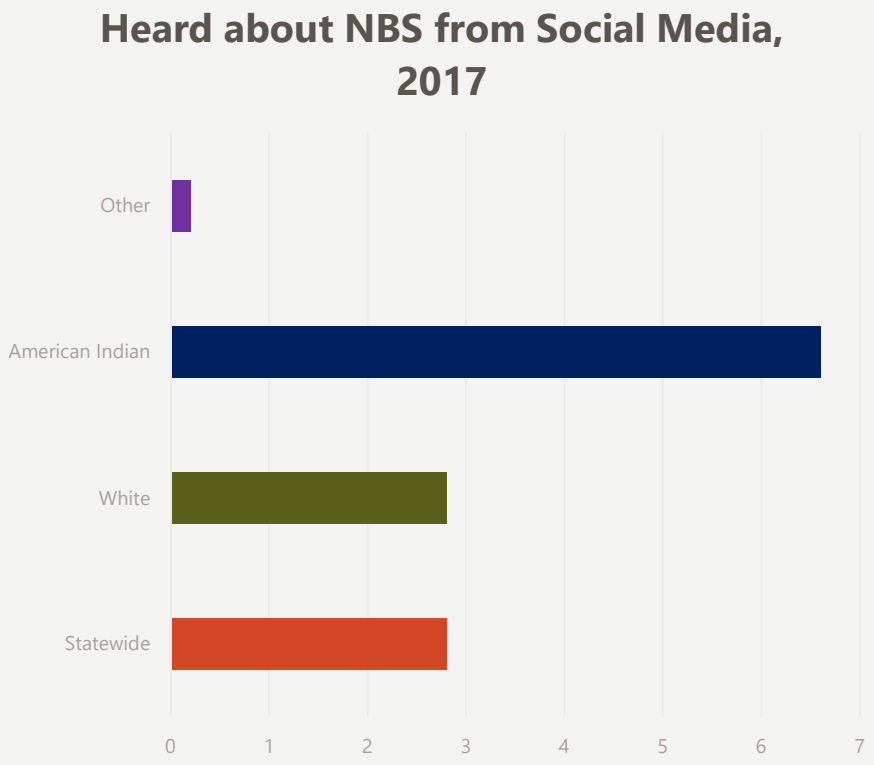
PRAMS 2017:NEWBORN SCREENING





PRAMS 2017: NEWBORN SCREENING





THANK YOU!

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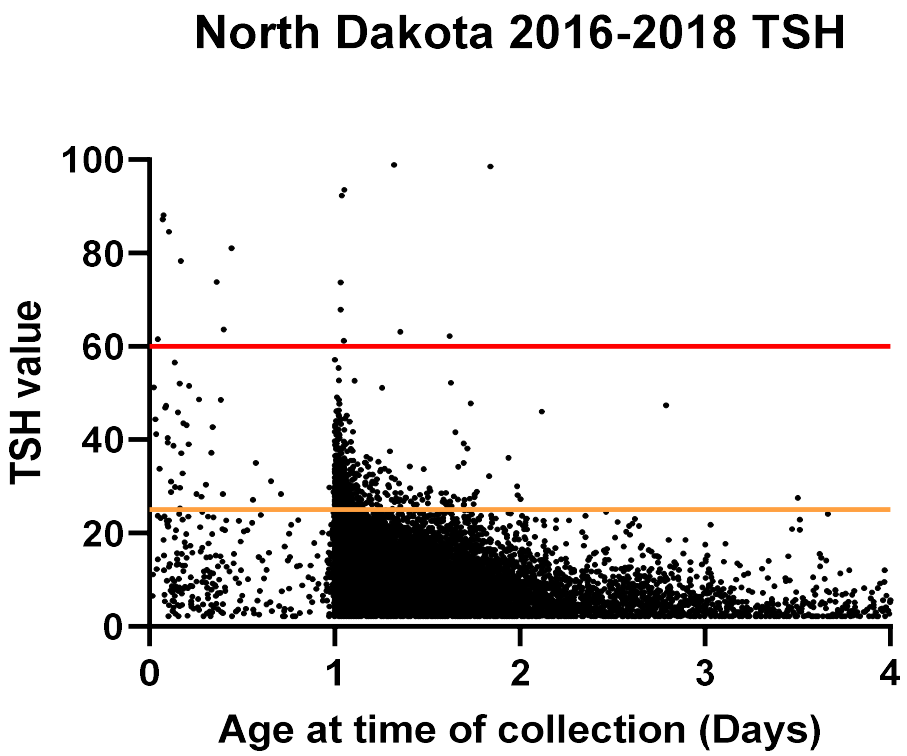
TSH UPDATE

PRESENTER: ANNE ATKINS, MPH (IOWA)

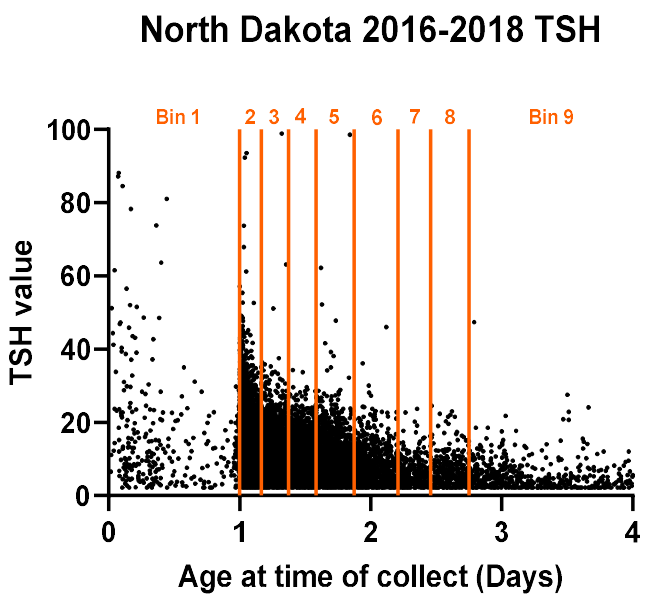
Age At Time Of Collection Cut-offs For Congenital Hypothyroidism In Newborn Screening

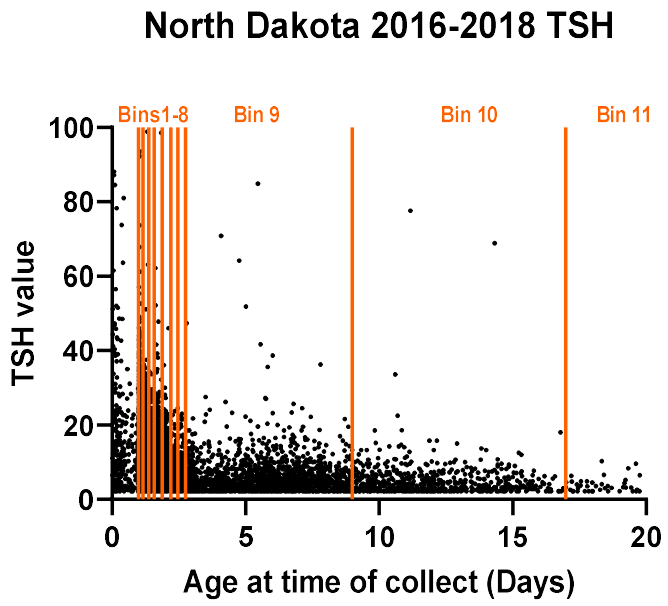
Background

- The current TSH algorithm:
  - > 25.0 mIU/ml = borderline result (a repeat specimen is requested)
  - >/= 60.0 mIU/ml as a presumptive positive result (diagnostic evaluation is requested)
- The current fixed cut-offs do not account for the rapid decrease in TSH following delivery



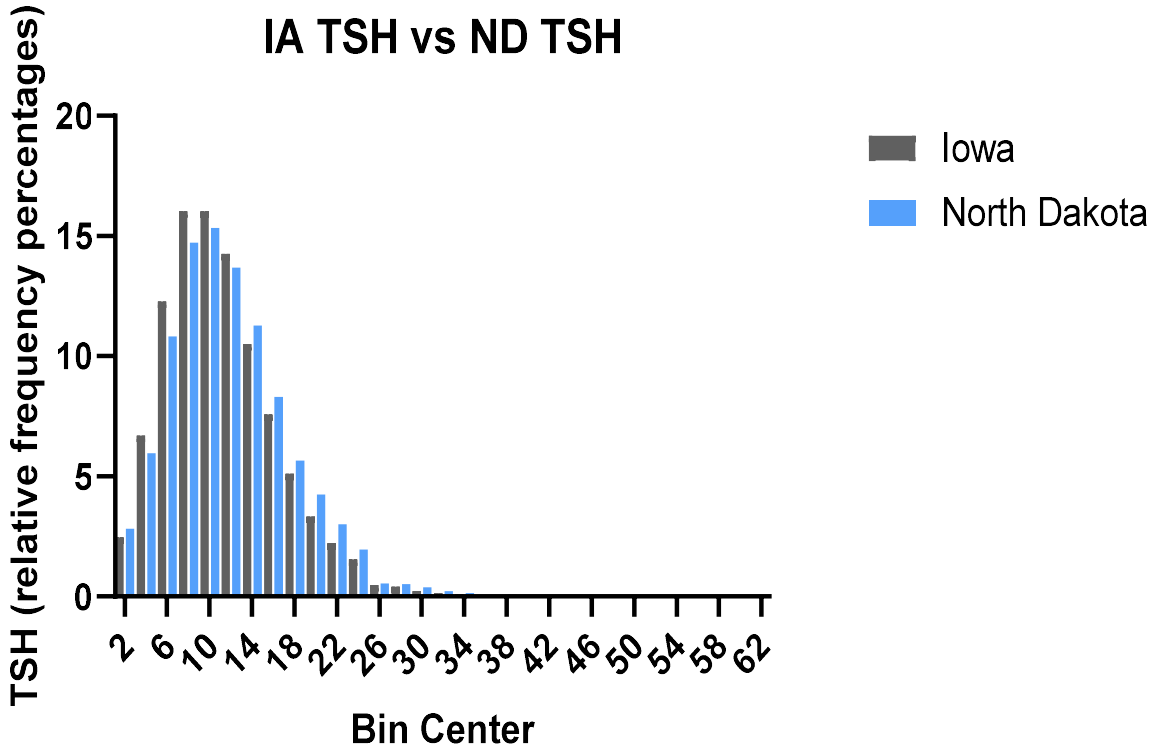
Proposal


North Dakota 2016-2018 TSH
Bin 1
2
3
4
5
6
7
8
Bin 9
TSH value
100
80
60
40
20
0
0
1
2
3
4
Age at time of collect (Days)


North Dakota 2016-2018 TSH
Bins1-8
Bin 9
Bin 10
Bin 11
TSH value
100
80
60
40
20
0
0
5
10
15
20
Age at time of collect (Days)

Distribution of Iowa TSH vs North Dakota TSH (2017-partial 2019)

The Iowa and North Dakota population TSH distributions are similar; therefore, we feel that it is appropriate to utilize the bins and cutoffs calculated from the Iowa data for the North Dakota population.



	Age at time of specimen collection	Borderline	Presumptive Positive
Bin 1	00:00 to 23:59	n/a	≥156
Bin 2	24:00 to 27:59	≥36	≥62
Bin 3	28:00 to 32:59	≥28	≥47
Bin 4	33:00 to 37:59	≥24	≥40
Bin 5	38:00 to 44:59	≥25	≥43
Bin 6	45:00 to 52:59	≥20	≥31
Bin 7	53:00 to 58:59	≥18	≥28
Bin 8	59:00 to 65:59	≥14	≥21
Bin 9	66:00 to 215:59	≥18	≥28
Bin 10	216:00 to 407:59	≥13	≥20
Bin 11	408:00 to End	≥11	≥16

Proposed borderline and presumptive positive TSH ranges (units: mIU/ml) within each age at time of collection bin.

Comparison of proposed binned TSH algorithm to current fixed cut-off algorithm

	Current Fixed Cutoffs					Proposed Cutoffs				
	2014	**2015**	**2016**	**2017**	**2018**	**2014**	**2015**	**2016**	**2017**	**2018**
n	13390	13271	13588	12792	12789	13390	13271	13588	12792	12789
bord	134	182	272	188	293	81	77	63	57	62
%bord	1.00%	1.37%	2.00%	1.47%	2.29%	0.60%	0.58%	0.46%	0.45%	0.48%
pp	12	12	20	9	11	15	21	23	9	14
%pp	0.09%	0.09%	0.15%	0.07%	0.09%	0.11%	0.16%	0.17%	0.07%	0.11%

Retrospective analyses

	2014		2015		2016		2017		2018	
	current	bins	current	bins	current	bins	current	bins	current	bins
NORMAL	0	0	0	0	2	2	0	0	0	1
BORDERLINE	2	1	2	1	4	4	1	1	3	0
PRESUMPTIVE POSITIVE	2	3	6	7	10	10	5	5	4	6

We assessed how the proposed age at time of collection cutoffs would change the laboratory call-out category for confirmed congenital hypothyroidism (CH) cases – as tracked by the Iowa Newborn Screening Follow-Up program. From 2014-2018 there were **41 confirmed CH cases** in North Dakota.

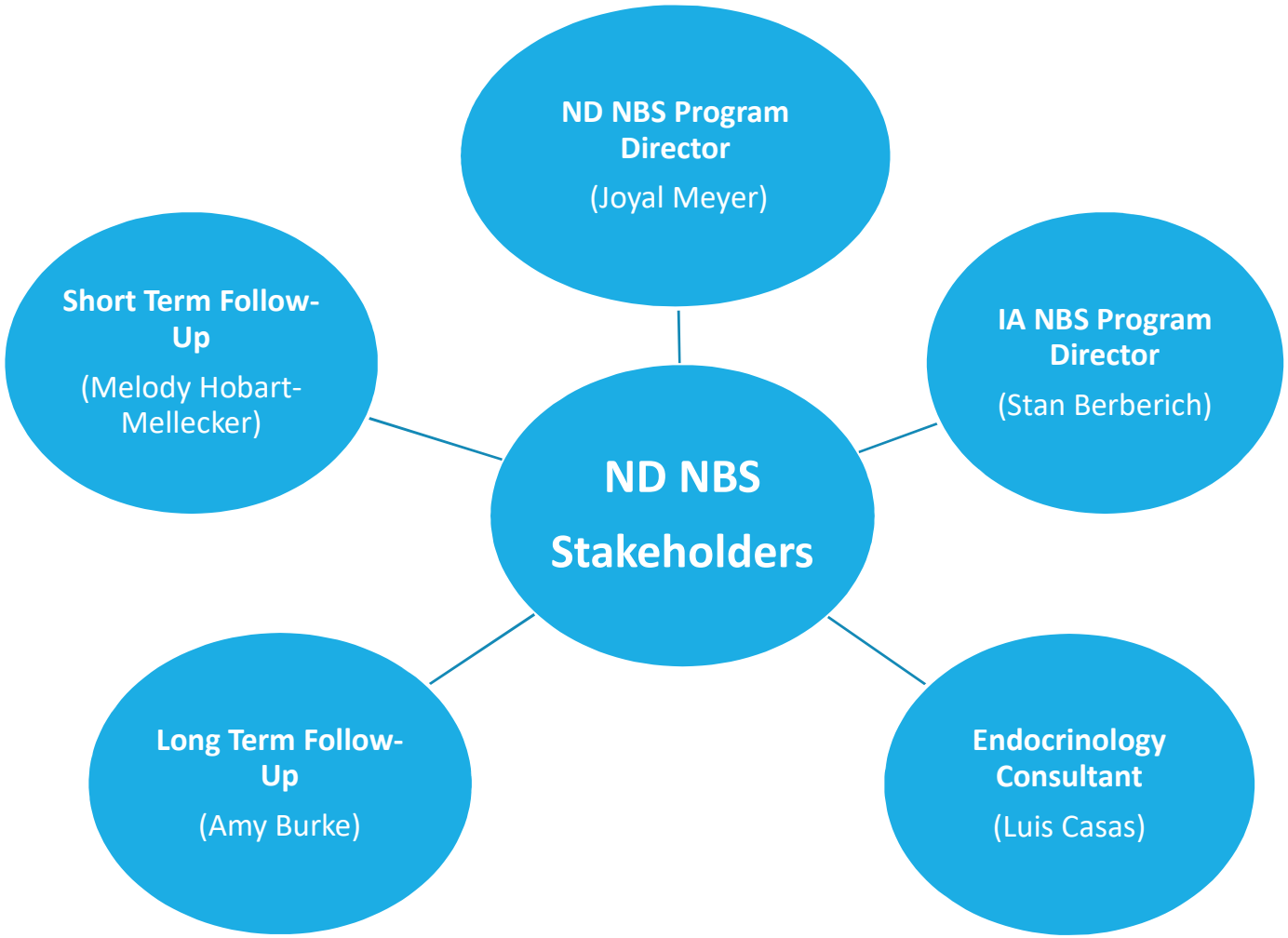
Retrospective analyses cont.

	2014	2015	2016	2017	2018
N to N	0	0	2	0	0
BORD to BORD	1	1	4	1	0
PP to PP	2	6	10	5	4
BORD to N	0	0	0	0	1
PP to BORD	0	0	0	0	0
N to BORD	0	0	0	0	0
BORD to PP	1	1	0	0	2

2018 North Dakota Numbers

	Current Cutoffs			Proposed TSH cutoffs			
	Initials	**Repeats**	**Total**	**Initials**	**Repeats**	**Total**	**Net Change**
Borderline	290	3	293	54	8	62	-231
Presumptive Positive	8	3	11	9	5	14	+3

Overall Impact to NBS Follow-Up


ND NBS Program Director
(Joyal Meyer)
Short Term Follow-Up
(Melody Hobart-Mellecker)
IA NBS Program Director
(Stan Berberich)
ND NBS Stakeholders
Long Term Follow-Up
(Amy Burke)
Endocrinology Consultant
(Luis Casas)

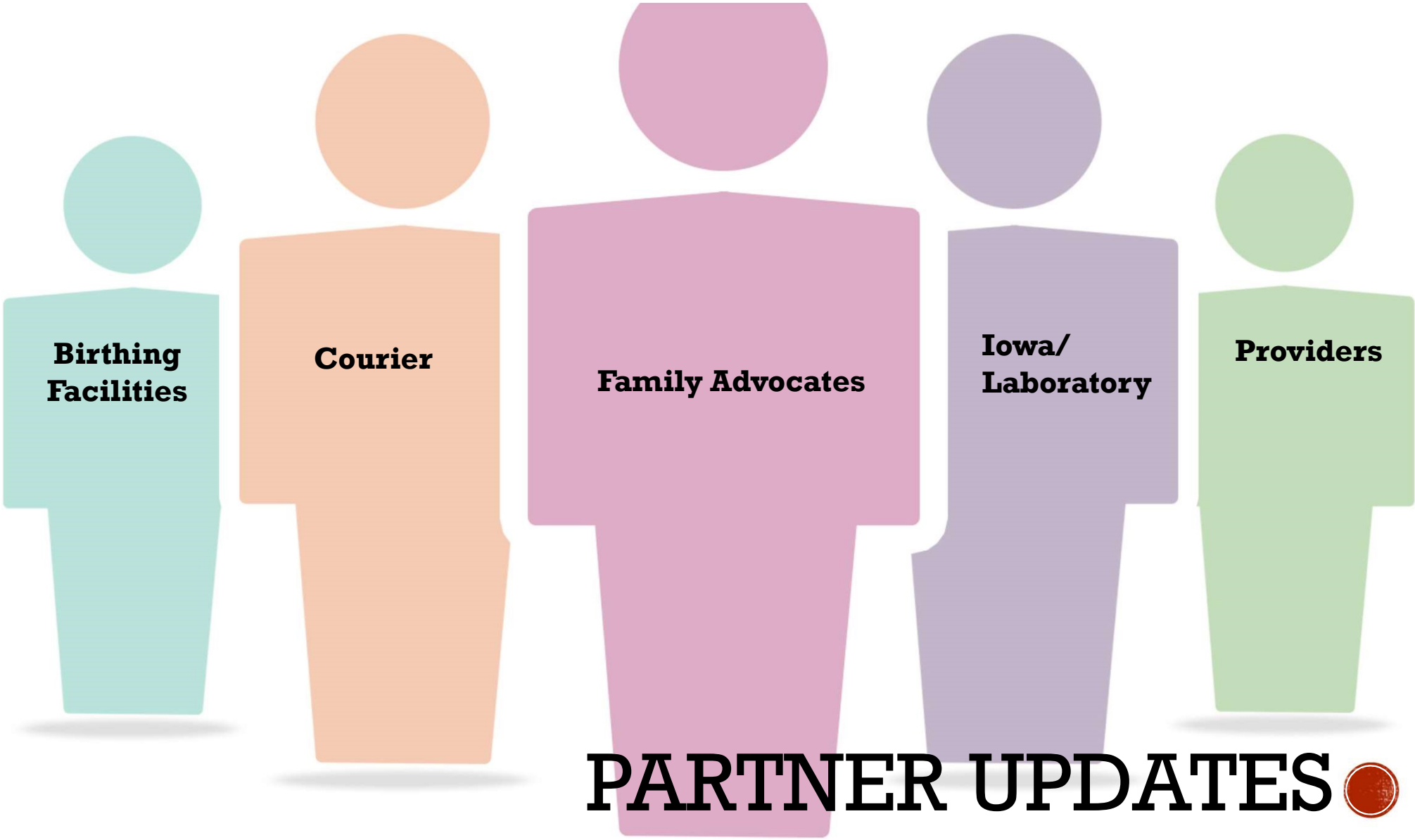
Next Steps

- LIMS and IT
- Laboratory changes
- Validation
- Education
  - Endocrinologists
  - Submitters
  - Peds community

Questions?

Retrospective Case – Bord to N

	TSH-NBS Initial	Rpt NBS?	TSH- NBS Rpt	Serum TSH – 1	Serum TSH – 2	Serum TSH - 3
1	31	NO		18.11		



PARTNER UPDATES

2020 ADVISORY MEETINGS

January

April

July

October

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Next Up:

THE LETTER-CASE PROJECT

Screening vs Diagnostic Tools for Prenatal Testing and Best Practices for Delivering Unexpected News

Presentation by: Dr A. Tobiasz